



KHDC3L gene

KH domain containing 3 like, subcortical maternal complex member

Normal Function

The *KHDC3L* gene provides instructions for making a protein that is involved in regulating gene activity (expression). The KHDC3L protein is thought to play a role in turning off (inactivating) genes based on which parent the copy of the gene came from, a phenomenon known as genomic imprinting. For most genes, both copies of the gene (one copy inherited from each parent) are active in all cells. For a small subset of genes, however, only one of the two copies is active; for some of these genes, the copy from the father is normally active, while for others, the copy from the mother is normally active. The KHDC3L protein is involved in imprinting multiple maternal genes in egg cells (oocytes) that contribute to development of the embryo, ensuring that the genes will be inactive; the corresponding paternal genes are active.

It is likely that the *KHDC3L* gene has additional roles in embryonic development; however, its exact functions are unclear.

Health Conditions Related to Genetic Changes

recurrent hydatidiform mole

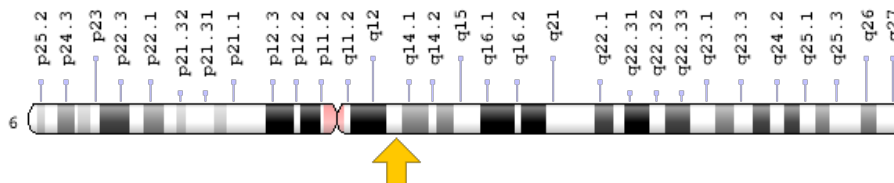
At least four mutations in the *KHDC3L* gene have been found to cause recurrent hydatidiform mole. A hydatidiform mole occurs early in pregnancy when an embryo does not fully develop and the placenta develops abnormally. The placenta, a solid structure in the uterus that normally provides nutrients to a growing fetus, is dysfunctional and appears as numerous small sacs, often described as resembling a bunch of grapes. There is a risk of the hydatidiform mole becoming cancerous. Women who have repeated instances of hydatidiform mole have a condition called recurrent hydatidiform mole. *KHDC3L* gene mutations are responsible for recurrent hydatidiform mole in 5 percent of women with this condition.

KHDC3L gene mutations result in the production of a protein with impaired function. As a result, the protein's role in the imprinting process is diminished, leading to the activation of many maternal genes that should not be expressed. The overexpression of multiple genes during embryonic development results in poor development of fetal and placental tissues characteristic of a hydatidiform mole. Because the *KHDC3L* gene mutations are present in all of a woman's cells, including oocytes, a hydatidiform mole will develop in each pregnancy that occurs with those egg cells.

Chromosomal Location

Cytogenetic Location: 6q13, which is the long (q) arm of chromosome 6 at position 13

Molecular Location: base pairs 73,362,677 to 73,364,175 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C6orf221
- ECAT1
- ES cell-associated transcript 1 protein
- HYDM2
- KH domain containing 3-like, subcortical maternal complex member
- KHDC3-like protein

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KHDC3L%5BTIAB%5D%29+OR+%28C6orf221%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- KHDC3-LIKE PROTEIN, SUBCORTICAL MATERNAL COMPLEX MEMBER
<http://omim.org/entry/611687>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=KHDC3L%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=33699
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/154288>
- UniProt
<http://www.uniprot.org/uniprot/Q587J8>

Sources for This Summary

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